

SUPPLEMENT TO THE PATIENT BOOKLETS FOR THE CALIFORNIA EXPANDED AFP SCREENING PROGRAM

**NTD/SLOS Only Screening: Second Trimester Screening For
Open Neural Tube Defects and Smith-Lemli-Opitz Syndrome**

The California Expanded AFP Screening Program offers an option for a second trimester blood test that screens only for Open Neural Tube Defects (NTD) and Smith-Lemli-Opitz Syndrome (SLOS). **This NTD/SLOS Only Screening is only for pregnant women who have already had CVS, Preimplantation Genetic Diagnosis for chromosome abnormalities, or First Trimester Screening AND who do not want additional screening for chromosome abnormalities.**

Chorionic Villus Sampling (CVS) is a test done on cells from the placenta between 10 and 12 weeks of pregnancy. It is a diagnostic test for chromosomal abnormalities in the fetus such as Down syndrome and Trisomy 18.

Preimplantation Genetic Diagnosis (PGD) is a test done on embryos before implantation during In Vitro Fertilization. When PGD is done to detect chromosome abnormalities, it can diagnose Down syndrome and Trisomy 18.

First Trimester Screening is a special ultrasound and blood test combination, usually done between 9 and 13 weeks of pregnancy. It is an early screening test for Down syndrome and Trisomy 18.

Since none of these tests are used to help detect NTD, Abdominal Wall Defects, or SLOS, there is a **Second Trimester blood test that screens just for these birth defects.** A woman who has had CVS, PGD for chromosome abnormalities, or First Trimester Screening can choose to have **NTD/SLOS Only Screening**, which does not include results for Down syndrome and Trisomy 18. Women who have had any of these first trimester tests should discuss their options for NTD/SLOS Only Screening with their clinician or a genetic counselor.

What Are The Birth Defects That May Be Detected With NTD/SLOS Only Screening?

Open Neural Tube Defects (NTD), including spina bifida and anencephaly, are birth defects caused by incomplete closing of the Neural Tube (spinal cord) during development. Spina bifida may lead to life long paralysis, but can be helped with special treatments. Anencephaly is caused by an undeveloped brain and is always fatal.

Abdominal Wall Defects (AWD) are problems involving the baby's abdomen and intestines. Surgery after birth often corrects the defect. Abdominal wall defects may be identified during testing for NTD.

SLOS (Smith-Lemli-Opitz Syndrome) is a problem with cholesterol production by the fetus. It causes mental retardation and can cause heart, hearing and other problems.

Continued on page 2

Patient Information Page

SUPPLEMENT TO THE PATIENT BOOKLETS FOR THE CALIFORNIA EXPANDED AFP SCREENING PROGRAM

NTD/SLOS Only Screening: Second Trimester Screening For Open Neural Tube Defects and Smith-Lemli-Opitz Syndrome

The Blood Test for NTD/SLOS Only Screening

The blood test for NTD/SLOS Only Screening is done between 15 and 20 weeks of pregnancy. It tests for levels of Alpha-fetoprotein (AFP), and the hormones human chorionic gonadotropin (hCG) and unconjugated estriol (uE3) which are produced when a woman is pregnant. The test results show what the chances are that the woman is carrying a fetus with an Open Neural Tube Defect or with SLOS. Test results for NTD/SLOS **do not include** results for chromosome abnormalities such as Down syndrome and Trisomy 18.

If the result is “*Screen Negative*,” follow-up tests through the Program are not considered necessary. If the result is “*Screen Positive*,” the Program will pay for Genetic Counseling and follow-up tests at a State-approved Prenatal Diagnosis Center. A “*Screen Positive*” result does not mean that there is a birth defect – it means that follow-up tests are indicated, because the risk for a birth defect is higher than usual. Most women with “*Screen Positive*” results will have normal follow-up tests and healthy babies.

Not every birth defect will be detected. The Expanded AFP Program detects about 80% of spina bifida, 97% of anencephaly, 85 % of abdominal wall defects, and 60% of SLOS.

The blood test costs \$162 and is covered by most insurance companies, once the patient has paid any co-payments or deductibles.

**If you choose to have NTD/SLOS Only Screening,
please sign the consent forms on the following pages.**

For the Patient who has had First Trimester Screening, CVS or Preimplantation Genetic Diagnosis

PATIENT'S COPY

Patient's name _____ ID# _____ (PLEASE PRINT)

CONSENT / REFUSAL FOR NTD/SLOS ONLY SCREENING THE CALIFORNIA EXPANDED AFP SCREENING PROGRAM

1. I have received the Expanded AFP Screening Program Basic or Choices Booklet. I have read or been informed that:
 - a. The purpose of "NTD/SLOS Only Screening" is to help detect fetuses with open neural tube defects, abdominal wall defects, and Smith-Lemli-Opitz syndrome. However, the Program cannot detect all such defects.
 - b. There may be other birth defects affecting the fetus that cannot be detected by this blood test.
 - c. I understand the "NTD/SLOS Only Screening" does not include screening results for Down syndrome or Trisomy 18.
 - d. If the result of the blood test is "**screen positive**", I will need to make a decision regarding follow-up testing. Authorized follow-up tests (at a State-approved Prenatal Diagnosis Center) are covered by the Program at no additional charge.
 - e. If the blood test result is "**screen negative**", the Program will not pay for any follow-up testing.
 - f. If the fetus is found to have a birth defect, the decision to continue or terminate the pregnancy will be entirely mine.
 - g. Participation in the California Expanded AFP Screening Program is voluntary. I can refuse any tests at any time.
2. I have been informed that a blood specimen for "NTD/SLOS Only Screening" must be drawn between 15 and 20 weeks of pregnancy.
3. I understand that the cost for "NTD/SLOS Only Screening" is \$162.
4. I have had my questions answered to my satisfaction.

Yes	<p>Instead of full Expanded AFP Screening, I request blood be drawn for "NTD/ SLOS Only Screening" which does not include results for Down syndrome or Trisomy 18.</p> <p>Signed _____ Date _____</p> <p>My blood has to be drawn between _____ and _____</p> <p>Month / Day /Year Month / Day /Year</p>
No	<p>I decline "NTD/SLOS only Screening".</p> <p>Signed _____ Date _____</p>

I understand that my blood specimen, and information obtained during the testing process, becomes the property of the California Department of Health Services. The Department or Department-approved scientific researchers may use them for program evaluation or research without identifying the person or persons for whom these results were obtained, unless I specifically prohibit such use in writing to the Genetic Disease Branch. All information is confidential.